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Conference Abstract

Two candidate genes for recurrent pregnancy loss and infertility: Could ZP3 and UPK3B give us new diagnostic and therapeutic approach?

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Abstract

Introduction:

Chromosomal indels are relatively common cytogenetic abnormalities. Nonetheless, clinical outcomes depend on the location, size and genes in deletion or duplication regions. The zona pellucida is an extracellular matrix that surrounds the oocyte and early embryo. It is composed primarily of three or four glycoproteins with various functions during fertilization and preimplantation development. The protein encoded by ZP3(Zona pellucida3) gene is a structural component of the zona pellucida and functions in primary binding and induction of the sperm acrosome reaction. UPK3B(Uroplakin 3B), a minor component of the apical plaques of mammalian urothelium that binds and dimerizes with uroplakin-1b(UPK1B), one of the major conserved urothelium membrane proteins. We herein report two cases presenting with the deletions encompassing POMZP3, UPK3B, ZP3, POM121 and POM121C genes.

Case1: 25-year-old female presented to our clinic with recurrent pregnancy loss. After clinical and cytogenetic evaluation, which all of them do not feature, she was diagnosed as the deletion of POMZP3 and UPK3B genes with the array-CGH platform. (Agilent SurePrintG3 HumanCGH 60K)

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Case2: Ten-week embrio of 34-year-old female, infertile for ten years before and this is her first pregnancy after IVF, revealed the deletion of POM121,POM121C and ZP3 genes with the same array-CGH platform.

Conclusion:

We have evaluated the deletion of two consecutive genes -UPK3B and ZP3- in the genome by array-CGH analysis. Early abortion or infertility due to triploidic, tetraploidic embryos or uniparental disomy, resulting in a change in the structure of the zona pellucida with the mutations of ZP3 gene, may occur. Also mutations of UPK3B gene may cause abortion or infertility due to endometrial origin with defective function of the urothelium membrane proteins. ICSI could be a good choice for ZP3 deficient infertile woman and if PGS choosen without ICSI, uniparental disomia should be excluded.

Keywords

Array-CGH, nfertility, Recurrent Pregnancy Loss , UPK3B gene, Zona Pellucida, ZP3 gene

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Conflicts of interest

The authors declare that there is no conflict of interests.